Understanding Genetics

WEATHERBYS

SCIENTIFIC

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While this booklet focuses on cattle as an example, this information is useful for all species.

DNA is essential to building all parts of all living things. Most people are familiar with the double helix structure discovered in 1953, but don't fully understand what it is or why we care about it. In the most basic terms, DNA is the building blocks of life. It is composed of 4 nucleotides, also called **bases**, adenine (A), cytosine (C), guanine (G), and thymine (T). These nucleotides are arranged on a sugar and phosphate backbone and when they are matched up, make up the double helix we have all become familiar with (Figure 1 A-C).
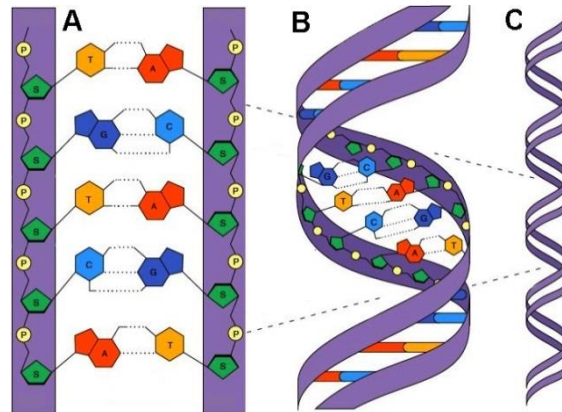

Figure 1:
A- If you were to zoom in, untwist the double helix, and flatten it out, it would look like this. Notice that the nucleotides in the centre are paired up, and the sugar and phosphate backbone, highlighted in purple, are on either side. This is what makes up every part of all living things.
B- Reassembling it into its double-helix structure and showing one twist of the helix.
C- Zooming out further to see multiple twists of the DNA helix.

The cattle genome has approximate 3 billion DNA bases, the same number as found in the human genome. To help store all this information DNA are packaged in **chromosomes**. These chromosomes can be broken down into 3 categories: autosomes, sex determining chromosomes (X and Y), and mitochondria. While most animals will share the same DNA code throughout the genome there are differences in the code which cause each animal to look and perform differently. Some of these differences in the DNA code (called **alleles** or mutations) can cause genetic diseases and a difference in phenotype (physical traits of the animal). Figure 2 is a pictorial depiction of alleles.

Over 100 of these differences that are known to cause a disease or trait are on the IDB SNP chip. When the alleles are described in the genetic defect and major gene booklet, you'll see A>T (or T>G, G>C,…) what this means is that most animals have the first base (A) and the allele we test for is the second base given (T). So for the G>T mutation in the SLC35A3 gene that causes Complex Vertebral Malformation (CVM) disease 'G' is the allele most animals have, and 'T' is the allele that causes the disease. Animals that have CVM will be homozygous (have 2 copies) for the 'T' allele.
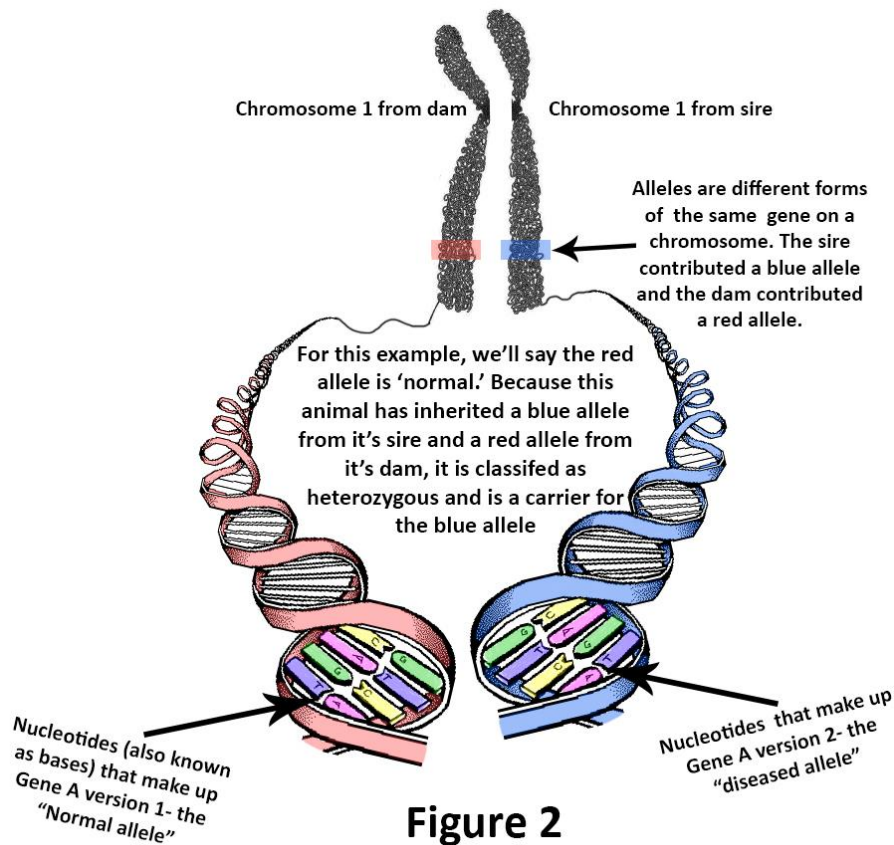
Figure 2

Figure 2: This image shows what happens in an animal produced from a sire and dam with different allele types for one gene. The dam has contributed a normal (red) allele while the male contributed a diseased (blue) allele to the offspring. This results in the offspring being a carrier of the diseased gene.

An animal's genetic disease status is described as Normal, Carrier, or Homozygous for X, where X is the disease name. These are defined below:

Normal= animal has 0 copies of the trait allele

Carrier = animal has 1 copy of the trait allele and may not show a phenotype

Homozygous = animal has 2 copies of the trait allele and will show the phenotype

A trait can be recessive, dominant, additive, or have interactions with other genes.

A **Recessive** trait means that an animal has to have 2 copies of the trait allele for the animal to be affected with the disease. Those with 1 or 0 copies have the normal phenotype. An example of this is the Complex Vertebral Malformation (CVM) disease allele. Animals with 2 copies of the CVM allele are aborted or born dead while those with 1 or 0 copies are normal.

A **Dominant** trait means that an animal with 1 or 2 copies of the allele will show the trait, while those with 0 copies have the normal phenotype. An example of this is the Polled allele. Animals with 1 or 2 copies of the Polled allele are polled while those with 0 copies of the allele have horns

An **Additive** trait means each copy of the trait allele increased the trait effect, and animal with 1 allele will have a phenotype that is between what is seen in an animal with 2 or 0 copies of the allele. An example of this is the Silver Dilutor 1 allele. Animals with 0 copies will be the breed's base colour such as Black, animals with 1 copy will be light grey, and animals with 2 copies will be white

Examples of the mating risk of having a genetic disease affected calf born when the dam and sire are normal, carrier, or homozygous for recessive or dominant disease alleles are shown below.

Recessive Disease Mating Risk

Animals with 2 copies of the trait allele have the trait phenotype (affected), those with 1 or 0 copies have the normal phenotype. A common way to write these different allele types is by using a capital letter (i.e. **A**) to designate the normal gene and a lower case letter to designate the affected gene (i.e. **a**). For these examples, we'll call an animal that is homozygous for the desirable trait, **normal**. This animal would have a gene designation as 'AA.' Animals that are **carriers** would get the designation 'Aa,' and we'll call the animals that have 2 copies of the diseased allele **homozygous** because they are homozygous for the diseased allele. These animals would have the designation 'aa.'

1. Mating a **normal** to **normal** results in a 0% chance of having an affected calf born

2. Mating a **normal** to a **carrier** results in a 0% chance of having an affected calf born

3. Mating a **normal** to **homozygous** results in a 0% chance of having an affected calf born

4. Mating a **carrier** to **carrier** results in a 25% chance of having an affected calf, a 50% chance of having a carrier calf, and a 25% chance of having a normal calf born.

5. Mating a **carrier** to **homozygous** results in a 50% chance of having a carrier calf and a 50% chance of having an affected calf born

6. Mating a **homozygous** to **homozygous** results in a 100% chance of having an affected calf born

A Punnett Square is a great way to graphically to express these matings. When making up a Punnett's square you make a 3X3 grid. The allele for the sire goes into the top middle and top right of the square, and the dam's allele type goes into the left bottom and left middle square (see below). Once set up, you carry the sire's alleles down and the dam's allele's across giving you the possible allele combinations if you mated those two animals. Punnett's square examples of mating 2 animals that are Normal (AA), Carrier (Aa), or Homozygous(aa) for a **RECESSIVE** trait are below:

Carrier sire X Carrier dam. How do we know they are both carriers? When looking at the gene designations at the top of the squares, they have one copy of the normal allele (**A**) and one copy of the affected allele (**a**).

	Sire	
Dam	**A**	**a**
A	AA Normal	Aa Carrier
a	aA Carrier	aa Affected

Carrier sire (Aa) X Normal dam (AA)

	Sire	
Dam	**A**	**a**
A	AA Normal	Aa Carrier
A	AA Normal	Aa Carrier

Carrier sire (Aa) X Homozygous dam (aa)

	Sire	
Dam	**A**	**a**
a	aA Carrier	aa Affected
a	aA Carrier	aa Affected

Dominant Disease Mating Risk

Animals with 1 or 2 copies of the trait allele have the phenotype (affected), those with 0 copies have the normal phenotype. This is just like above, but this time if a calf gets ANY copies of the affected allele (a), it will be affected.

1. Mating a **normal** to **normal** results in a 0% chance of having an affected calf born

2. Mating a **normal** to **carrier** results in 50% chance of having a normal calf and a 50% chance of having a affected calf born

3. Mating a **normal** to **homozygous** results in a 100% chance of having a carrier calf born that is affected with the trait

4. Mating a **carrier** to **carrier** results in a 75% chance that the calf born will be affected and a 25% chance that it will be normal.

5. Mating a **carrier** to **homozygous** results in a 100% chance of having an affected calf born.

6. Mating a **homozygous** to **homozygous** results in a 100% chance of having an affected homozygous calf born

Punnett Square examples of mating 2 animals that are Normal, Carrier, or Homozygous for a **DOMINANT** trait are below:

Carrier sire (Aa) X Carrier dam (Aa)

	Sire	
	A	**a**
A	AA Normal	Aa Affected
a	aA Affected	aa Affected

(Dam)

Carrier sire (Aa) X Normal dam (AA)

	Sire	
	A	**a**
A	AA Normal	Aa Affected
A	AA Normal	Aa Affected

(Dam)

Carrier sire (Aa) X Homozygous dam (aa)

		Sire	
		A	a
Dam	a	aA Affected	aa Affected
	a	aA Affected	aa Affected